DDX11 gene

DEAD/H-box helicase 11

Normal Function

The *DDX11* gene provides instructions for making an enzyme called ChlR1, which functions as a helicase. Helicases are enzymes that attach (bind) to DNA and temporarily unwind the two spiral strands (double helix) of the DNA molecule so it can be copied (replicated) in preparation for cell division. ChlR1 is also involved in repairing any mistakes that are made when DNA is copied. In addition, ChlR1 is involved in other processes leading up to cell division. After replication, the DNA from each chromosome is arranged into two identical structures, called sister chromatids, which the ChlR1 enzyme helps to keep together until they are ready to separate into individual cells. This enzyme also ensures proper separation of chromatids during cell division. By helping repair mistakes in DNA and ensuring proper DNA replication, the ChlR1 enzyme plays a role in maintaining the stability of a cell's genetic information.

Health Conditions Related to Genetic Changes

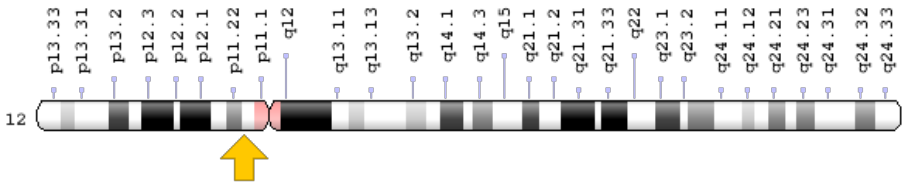
Warsaw breakage syndrome

At least three mutations in the *DDX11* gene have been found to cause Warsaw breakage syndrome. This condition causes multiple abnormalities that may include impaired growth, distinctive facial features, hearing loss, and heart malformations. The mutations that cause Warsaw breakage syndrome severely reduce or completely eliminate ChlR1 enzyme activity. As a result, the enzyme cannot bind to DNA and cannot unwind the DNA strands to help with DNA replication and repair. A lack of functional ChlR1 impairs cell division and leads to an accumulation of DNA damage. This DNA damage can appear as breaks in the DNA, giving the condition its name. It is unclear how these problems in DNA maintenance lead to the specific abnormalities characteristic of Warsaw breakage syndrome.

Chromosomal Location

Cytogenetic Location: 12p11.21, which is the short (p) arm of chromosome 12 at position 11.21

Molecular Location: base pairs 31,073,574 to 31,104,799 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CHL1-related helicase gene-1
- CHL1-related protein 1
- CHLR1
- DDX11_HUMAN
- DEAD/H (Asp-Glu-Ala-Asp/His) box helicase 11
- DEAD/H (Asp-Glu-Ala-Asp/His) box polypeptide 11 (CHL1-like helicase homolog, S. cerevisiae)
- DEAD/H box protein 11
- hCHLR1
- keratinocyte growth factor-regulated gene 2 protein
- KRG-2
- KRG2
- probable ATP-dependent RNA helicase DDX11

Additional Information & Resources

Educational Resources

- Genomes (second edition, 2002): The Replication Process
  https://www.ncbi.nlm.nih.gov/books/NBK21113/#A8121
- The Cell: A Molecular Approach: Action of Helicases and Single-Stranded DNA-Binding Proteins (figure)
  https://www.ncbi.nlm.nih.gov/books/NBK9940/figure/A783/?report=objectonly

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28DDX11%5BTIAB%5D%29+OR+%28CHLR1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- DEAD/H BOX 11
  http://omim.org/entry/601150

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DDX11.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DDX11%5Bgene%5D
- HGNC Gene Family: DEAD-box helicases
  http://www.genenames.org/cgi-bin/genefamilies/set/499
- HGNC Gene Family: DNA helicases
  http://www.genenames.org/cgi-bin/genefamilies/set/1167
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2736
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1663
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